A FAMILY FORM OF IDIOCY, GENERALLY FATAL, ASSOCIATED WITH EARLY BLINDNESS. (AMAUROTIC FAMILY IDIOCY).

Dr. B. SACHS, in his Inaugural Address as President of the Society, discussed this subject. He said that in July, 1887, at a meeting of the American Neurological Association, he had reported a case of arrested cerebral development with special reference to its morbid pathology. The child had been normal at birth, but at the age of three months there had been a cessation of mental progress, and the limbs had become weak. Vision had been gradually lost, and after awhile the child had become marasmic. It died at the age of two years. At first the case appeared to be an isolated instance of idiocy associated with amaurosis. Four years later, another similar case occurred in the same family, and at its death, its brain presented changes identical with those found in the brain of the first child. During the past few years the importance of this disease as a family affection had grown upon him. Attention had been directed to it in various articles, but up to this time it had attracted but little notice. In 1891, a child had been brought to him who had been normal up to six months of age, and in whom there had been a cessation or retrogression of the mental and physical development. There were present idiocy, spastic paraplegia, a mere perception of light, and a decided tendency to marasmus. The mother of this child had three other children who had remained normal up to three months of age. All three had died before the age of two years. At present there was in the Montefiore Home, New York City, a child of four years, who was completely idiotic and blind. Early in childhood the blindness had first developed. A sister had also developed early blindness.

Through the kindness of Dr. H. Knapp the speaker said he had been able to review the ophthalmological literature on this subject. In 1881, in the Transactions of the Ophthalmological Society of the United Kingdom, Vol. I., Tay described a case presenting symmetrical changes in the region of the yellow spot in each eye of an infant of twelve months. There was also weakness, but no paralysis in this case. The optic disks were at first apparently healthy, but subsequently a large white patch, having a brownish centre, appeared near the yellow spot. It did not look like a hemorrhage or a deposit of pigment, but presented rather the appearance seen in cases of embolism of the central artery of the retina. Five months later, in addition to the changes just described, the disks were found atrophic. Three similar cases had occurred in the same family. All of these had presented the same ocular symptoms, and all had died under the age of three years. In 1885, Dr. Knapp had reported on the ocular condition of one of the children already referred to by the speaker. Dr. Sachs said he had collected nineteen undoubted cases of this disease—five boys, eleven girls, and three in which the sex was not stated. Eight of these cases had come under his own notice. The first patient coming under his observation was a little girl, born at full term of healthy parents. She apparently did well up to three months of age, when she developed nystagmus, weakness and inability to use any of the muscles, and with this there was a cessation of mental growth. Dr. Knapp examined the child and found vibratory nystagmus; contracted pupils; the fovea centralis of a cherry-red color and surrounded by an intense grayish-white opacity. At a subsequent examination the child was found to be totally blind; the optic nerves were completely atrophied, and the disks perfectly white, with scarcely a trace of blood vessels. There was marked sensibility to auditory and tactile impressions. There was no evidence of inherited or acquired syphilis, or of rachitis. In the same family the next child was a healthy vigorous boy, but the third child presented exactly the same symptoms as the first, and the morbid findings in the brain were the same. Special care had been taken in rearing this child, yet there had been a cessation of mental development at the age of eight months. It had been seen by the speaker when thirteen months old. At this time, the paresis was marked and spastic. Six months later, sight and hearing had been completely lost, and at the age of twenty months—four months earlier than the first child—it died in extreme narasmus. In August, 1891, he had seen a child who at birth was said to have weighed twelve pounds. The child had been nursed, and had done well up to six months. At thirteen months, examination showed the fontanelles open; the child unable to sit or stand or walk; the eyes divergent, but no nystagmus. Dr. Kingdon had reported a case seen by him in the Children's Hospital at Nottingham. The pupils in this case were equal, and reacted readily to light. The optic disks were clear and pale; there was no venous engorgement, and no hemorrhages. At the yellow spot, covering a space nearly twice the size of the optic papilla was a grayish-white patch, with few vessels. The fovea was of a cherry-red color. This child died when twelve months old. Six years before this, a boy in the same family had wasted away and died when two years old; and still another child born after the one here referred to, was afflicted with the same disease.

The prominent features in the cases under consider ation, the speaker said, might be briefly stated as follows: (1) Cessation of mental development and idiocy at the age of a few months; (2) paresis of the greater part of the body, either flaccid or spastic; (3) the reflexes may be deficient or increased; (4) diminution of vision terminating in absolute blindness with changes in the macula and later an optic nerve atrophy; (5) marasmus and a fatal termination at about two years of age; and (6) the occurrence of the affection in several members of the same family. Nystagmus, strabismus, and hyperacuity of hearing had been observed in most of the cases.

Out of the nineteen recorded cases, the same ophthalmoscopic appearances had been found in fourteen; hence these changes were the most important of all the signs. Post mortem examinations in these cases showed no evidence of any previous encephalitic process, and no proliferation in the walls of the blood vessels. The most marked changes were in the pyramidal cells of the cortex. In another case there were certain areas of degeneration found in the cord, the exact nature of which had not been determined. Clinical observation indicated that the primary etiological factors were, marked neurotic taint, blood relationship of the parents and traumatism in the mother during pregnancy. The condition had been so far almost exclusively observed among the Hebrews. In no case had there been any evidence of syphilitic taint. Dr. Sachs said that "agenesis corticalis' would seem to be a fitting term to describe this condition, but possibly a more generally useful one in the present state of our knowledge would be "amaurotic family idiocy."

Dr. KOLLER: I have listened with profound interest to the paper of Dr. Sachs, describing this apparently typical disease. I had communicated to him the two cases which I had observed but had failed to connect them with those described by Warren, Tay, Kingdon, Goldzieher, Magnus, Wadsworth, and Knapp. Indeed they present many features that would justify their being incorporated in this group, in fact the similarity over-balances the differences, which nevertheless exist.

On June 18, 1894, Mary L., then two years old, was brought to me for examination by her mother. When the child was three months old it was noticed that she did not use her eyes as other children of that age do. There was lateral nystagmus present, which according to the history given had developed in the first few months of the child's life. The ophthalmoscopic examination, difficult on account of the floating image, showed the discs to be in a congested state; besides I find in my record the entry, that apparently there is perception of light. There was nothing that struck me as unusual in the general condition of the child. The case was considered by me to be one of optic neuritis from an unknown cause. I did not see the child again until two years later, when an almost identical condition in an infant sister came under my observation, excited my interest and caused me to re-examine the first child. The general conditions of muscular debility and dementia had meanwhile developed or progressed as described by Dr. Sachs. At this examination I entered "Apparently perception of light, as child turns towards gas flame. Lateral nystagmus, not constant. Pupils of medium width, become slowly narrower when turning towards window, no prompt reaction. Both discs yellowish, discolored, present the conditions of atrophy as observed in retinitis pigmentosa. Veins dilated, arteries thinner. Dilated condition of veins may be due to gorging by screaming and struggling of the child while being held for examination. A more recent examination by Dr. Koller revealed the following condition:

Discs atrophic; yellowish discoloration similiar to appearance in retinitis pigmentosa. Retina atrophic; very thin chorioidal vessel visible through retina. In the region of the yellow spot a slight veil-like milkyblue opacity, gradually fading into the surrounding retina. In the centre of this opacity, at the site of the fovea centralis, a cherry-red patch, not very dark, a little smaller than the papilla, with ill-defined outlines.

On January 8, 1896, the infant sister, Hattie L., was presented for examination. The child was two months old, had lateral nystagmus. The mother thought the child was blind, but agreed with me that perception of light was present. The pupils of medium width, become slowly narrower when exposed to strong light. Both these children have been found to exhibit changes in the region of the macula, similar to those observed in the cases described by other oculists.

Dr. W. A. HOLDEN said that he had carefully examined one of the cases on record—that reported by Dr. Carter. The peculiar appearance about the macula and the progressive white atrophy of the disk would certainly attract the attention of the ophthalmologist. The appearance was exactly that seen in cases of stoppage of the arterial circulation of the retina when there is general œdema. It was hard to explain the appearance presented.